

A minor (or soft) marker is a difference that is seen on ultrasound that does not cause any health concerns itself, but increases the chance for certain underlying health conditions. Most of the time, a minor marker is a slight variation in development that is not linked to an underlying health issue.

For example, shorter than average femur (thigh) bones are considered a minor marker that increases the chance for <u>Down syndrome</u>. This is because many babies with <u>Down syndrome</u> have shorter than average thigh bones. But there are also many babies who do NOT have <u>Down syndrome</u> but DO have shorter than average thigh bones. There are also some babies with Down syndrome that have average thigh bones. If a minor marker is found on your ultrasound, it is important to talk with your doctor and/or genetic counselor to understand what that marker means for your pregnancy.

What are some other minor markers that may be seen in the 2nd trimester?

- Shortened or Absent Nasal Bone
- Choroid Plexus Cyst
- Echogenic Intracardiac Focus
- Increased thickness of the nuchal fold
- Renal Pyelectatsis/Hydronephrosis (enlarged kidneys)
- Short femur bones
- Short humerus bones
- Single umbilical artery (SUA), also called a two vessel cord

What if a minor marker or multiple markers are seen on my ultrasound?

You should be talk with a medical professional that can provide you more information about what these findings may mean for your baby. Depending on what marker(s) is seen, your doctor may offer you a screening test, such as cell-free DNA, or a diagnostic testing, such as amniocentesis, to look for certain genetic and chromosomal conditions like Down syndrome. It is always a personal choice whether or not to undergo any prenatal testing.

Depending on the markers that are seen on the ultrasound, it may be recommended that you have a follow-up specialized ultrasound to more carefully evaluate the baby's heart, called a fetal echocardiogram. Babies with chromosome conditions including Down syndrome have a higher chance for a heart defect. If an ultrasound increases the chance for the pregnancy to have a chromosome condition, it may be important in planning your delivery to determine if the baby is likely to have a heart defect.



Click $\underline{\text{here}}$ to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.